

***PRSS1* IVS2+1G>A c.200+1G>A**

Citation

- Chen JM, Le Maréchal C, Lucas D, Raguénès O, Férec C. (2003) **"Loss of function" mutations in the cationic trypsinogen gene (PRSS1) may act as a protective factor against pancreatitis.** Mol Genet Metab 79, 67-70
Variant was found in a subject with chronic alcoholism but without pancreatitis.

Functional studies

Note that variant is a splice site mutation.